AMN gene

amnion associated transmembrane protein

Normal Function

The *AMN* gene provides instructions for making a protein called amnionless. This protein is involved in the uptake of vitamin B12 (also called cobalamin) from food. Vitamin B12, which cannot be made in the body and can only be obtained from food, is essential for the formation of DNA and proteins, the production of cellular energy, and the breakdown of fats. This vitamin is involved in the formation of red blood cells and maintenance of the brain and spinal cord (central nervous system).

The amnionless protein is primarily found embedded in the outer membrane of kidney cells and cells that line the small intestine. Amnionless attaches (binds) to another protein called cubilin, anchoring cubilin to the cell membrane. Cubilin can interact with molecules and proteins passing through the intestine or kidneys. During digestion, vitamin B12 is released from food. As the vitamin passes through the small intestine, cubilin binds to it. Amnionless helps transfer the cubilin-vitamin B12 complex into the intestinal cell. From there, the vitamin is released into the blood and transported throughout the body. In the kidneys, amnionless and cubilin are involved in the reabsorption of certain proteins that would otherwise be released in urine.

Health Conditions Related to Genetic Changes

Imerslund-Gräsbeck syndrome

At least 30 mutations in the *AMN* gene have been found to cause Imerslund-Gräsbeck syndrome. This condition is characterized by low levels of vitamin B12 in the body, which leads to a blood disorder known as megaloblastic anemia. About half of affected individuals also have excess protein in their urine (proteinuria), and some have neurological problems.

AMN gene mutations that cause Imerslund-Gräsbeck syndrome reduce the amount or function of the amnionless protein. Without amnionless acting as an anchor, cubilin is not attached to cells in the small intestine or kidneys and cannot bind to vitamin B12 and other molecules and proteins needed in the body. As a result, instead of being taken into intestinal cells, vitamin B12 is released from the body. A shortage of this essential vitamin impairs the proper development of red blood cells, leading to megaloblastic anemia. Low levels of vitamin B12 can also affect the central nervous

system, causing neurological problems. In addition, without amnionless function in the kidneys, proteins are not reabsorbed into the body and are instead released in the urine, leading to proteinuria.

Other Names for This Gene

- amnionless
- amnionless homolog
- PRO1028
- protein amnionless
- protein amnionless precursor
- visceral endoderm-specific type 1 transmembrane protein

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=81693[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMN%5BTIAB%5D%29+OR+%28amnionless%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- AMNION-ASSOCIATED TRANSMEMBRANE PROTEIN; AMN (https://omim.org/entry/605799)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/81693)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AMN[gene])

References

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Genomic Location

The *AMN* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

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